



NF1 gene

neurofibromin 1

Normal Function

The *NF1* gene provides instructions for making a protein called neurofibromin. This protein is produced in many types of cells, including nerve cells and specialized cells called oligodendrocytes and Schwann cells that surround nerves. These specialized cells form myelin sheaths, which are the fatty coverings that insulate and protect certain nerve cells.

Neurofibromin acts as a tumor suppressor protein. Tumor suppressors normally prevent cells from growing and dividing too rapidly or in an uncontrolled way. This protein appears to prevent cell overgrowth by turning off another protein (called ras) that stimulates cell growth and division. Other potential functions for neurofibromin are under investigation.

Health Conditions Related to Genetic Changes

neurofibromatosis type 1

More than 1,000 *NF1* mutations that cause neurofibromatosis type 1 have been identified. Most of these mutations are unique to a particular family. Many *NF1* mutations result in the production of an extremely short version of neurofibromin. This shortened protein cannot perform its normal job of inhibiting cell division. When mutations occur in both copies of the *NF1* gene in Schwann cells, the resulting loss of neurofibromin allows noncancerous tumors called neurofibromas to form. Research indicates that the formation of neurofibromas requires the interaction of Schwann cells with other cells, including mast cells. Mast cells are normally involved in wound healing and tissue repair.

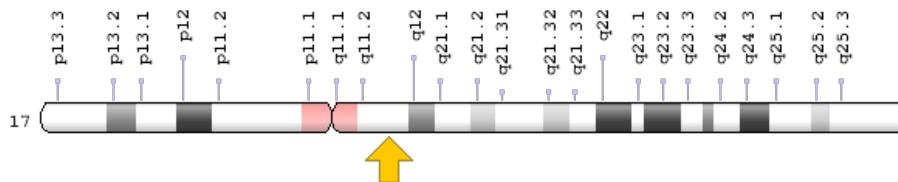
cancers

In rare cases, inactivation of one copy of the *NF1* gene in each cell increases the risk of developing juvenile myelomonocytic leukemia (JMML). Juvenile myelomonocytic leukemia is cancer of blood-forming tissue that usually occurs in children younger than 2. This condition causes the bone marrow to make an excessive number of immature white blood cells that cannot carry out their normal infection-fighting functions. These abnormal cells can build up in the blood and bone marrow, leaving less room for healthy white blood cells, red blood cells, and platelets. Children affected by this disorder may experience fatigue, fever, and easy bleeding or bruising.

Chromosomal Location

Cytogenetic Location: 17q11.2, which is the long (q) arm of chromosome 17 at position 11.2

Molecular Location: base pairs 31,094,927 to 31,377,677 on chromosome 17 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- Neurofibromatosis-related protein NF-1
- Neurofibromatosis Type 1 Protein
- neurofibromin 1 (neurofibromatosis, von Recklinghausen disease, Watson disease)
- NF1-GAP-Related Protein
- NF1 GRP
- NF1 Protein
- NF1_HUMAN

Additional Information & Resources

Educational Resources

- Cancer Medicine (sixth edition, 2003): NF1 and NF2 Genes
<https://www.ncbi.nlm.nih.gov/books/NBK12681/>
- National Cancer Institute Topic: Childhood Acute Myeloid Leukemia and Other Myeloid Malignancies
<https://www.cancer.gov/types/leukemia/patient/child-aml-treatment-pdq>

GeneReviews

- Neurofibromatosis 1
<https://www.ncbi.nlm.nih.gov/books/NBK1109>

Genetic Testing Registry

- GTR: Genetic tests for NF1
<https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=4763%5Bgeneid%5D>

Scientific articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28NF1%5BTI%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D>

OMIM

- JUVENILE MYELOMONOCYTIC LEUKEMIA
<http://omim.org/entry/607785>
- NEUROFIBROMIN 1
<http://omim.org/entry/613113>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
<http://atlasgeneticsoncology.org/Genes/NF1ID134.html>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=7765
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/4763>
- UniProt
<http://www.uniprot.org/uniprot/P21359>

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